Molecular genetic study of a severe growth hormone deficiency in a Chilean family Estudio genético molecular de una deficiencia severa de hormona del crecimento en una familia chilena.

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The first and so far the only form of isolated growth hormone deficiency to accede to molecular genetic analysis is IGHD 1A, which has an autosomal recessive mode of inheritance. Several sizes of growth hormone gene deletions, ranging from 6.7 to 8.5 Kb are responsible for IGHD 1A. It has been reported that children with IGHD 1A have an initial good response to growth hormone treatment, followed by growth failure associated with the development of blocking antibodies against growth hormone. The effectiveness of treatment has been associated to the size of the gene deletion. We performed a molecular genetic analysis of the growth hormone locus, using Southern Blot and PCR techniques, to a 12 years old girl who fulfilled the criteria for IGHD 1A and her closer relatives. The results showed a 7 Kb deletion for the growth hormone gene in the girl and one of her brothers.